



## From POF to POI: Evolution of a Term

About 10 years ago we began seeing the first reports of increased rates of infertility and early menopause in women with FMR1 gene premutations. Since then, we have learned that approximately 22 percent of premutation carriers have some degree of ovarian dysfunction. Many of these carriers were diagnosed with “POF” (premature ovarian failure), meaning their ovaries were functioning similarly to those of a woman in menopause. However, over the last few years, specialists in the field have become increasingly uncomfortable with referring to the ovarian complications in FMR1 carriers as “POF.” There are several reasons for this evolution in their thinking.

First, the term “POF” does not include the full spectrum of clinical ovarian symptoms in women with premutations. Some carriers merely have reduced fertility or irregular cycles, which are milder symptoms than complete premature ovarian failure. Additionally, the word “failure” suggests that it is impossible for women to get pregnant, but that has turned out not to be true. About 5 percent of women with “POF” do manage to conceive a child after receiving this diagnosis.

For these reasons, reproductive specialists have decided that *a more accurate term for this condition is primary ovarian insufficiency, or “POI.”*

“Primary” means that the disorder originates in the ovary, as opposed to some other organ or hormonal source, such as the brain.

“Insufficiency” means that the ovaries are not functioning to their normal capacity. This differs from “failure,” which indicates a complete shutting down of the ovary.

Recommendations to replace the term “POF” with “POI” have recently been published in infertility medical journals. This trend is occurring outside the field of Fragile X, among scientists and clinicians working with populations of women who have POI for reasons other than an FX premutation.

Eventually, POF may be completely replaced by POI in the entire medical community and the patient information materials that flow from its research. That is why, after careful deliberation and much discussion with those most knowledgeable in the field, we have decided to use this more scientifically accurate term in all of our own materials at The National Fragile X Foundation. In the coming months, readers will see the change reflected in this journal, our website, and in the educational literature we produce for families. And since the NFXF will be addressing POI only as a manifestation of Fragile X, it will be referred to as “fragile X-associated primary ovarian insufficiency” (FXPOI).

We trust that this change is easily understandable, and reflective of the ever-evolving knowledge we are gaining almost daily in this relatively new and dynamic field of Fragile X studies. ☞

*The author is a certified genetics counselor and the staff genetic specialist at the NFXF. Email: liane@FragileX.org*

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